BEST AVAILABLE IMAGES

Defective images within this document are accurate representations of the original documents submitted by the applicant.

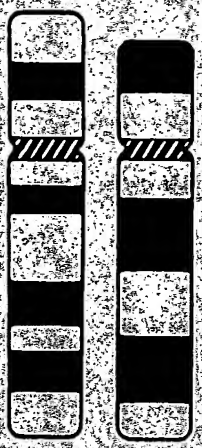
Defects in the images may include (but are not limited to):

- BLACK BORDERS
- TEXT CUT OFF AT TOP, BOTTOM OR SIDES
- FADED TEXT
- ILLEGIBLE TEXT
- SKEWED/SLANTED IMAGES
- COLORED PHOTOS
- BLACK OR VERY BLACK AND WHITE DARK PHOTOS
- GRAY SCALE DOCUMENTS

IMAGES ARE BEST AVAILABLE COPY.

As rescanning documents *will not* correct images, please do not report the images to the Image Problem Mailbox.

CHROMOSOMAL VARIATION IN MAN



A Catalog of Chromosomal Variants and Anomalies

7th Edition

Digamber S. Borgaonkar

CHROMOSOMAL VARIATION IN MAN

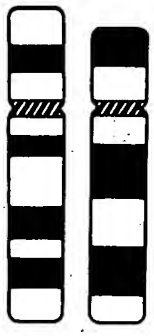
A Catalog of Chromosomal Variants and Anomalies

7th Edition

CHROMOSOMAL VARIATION IN MAN

A Catalog of Chromosomal Variants and Anomalies

7th Edition



Digamber S. Borgaonkar, Ph.D.

Director, Cytogenetics Laboratory, Dr. Margaret I. Handy Chair in Human Genetics,
Department of Pathology and Laboratory Medicine, Medical Center of Delaware, Newark, Delaware;
Adjunct Professor, School of Life and Health Sciences, University of Delaware;
and Research Professor in Genetics, Thomas Jefferson University

WILEY-LISS

A JOHN WILEY & SONS, INC., PUBLICATION
New York • Chichester • Brisbane • Toronto • Singapore

Address All Inquiries to the Publisher
Wiley-Liss, Inc., 605 Third Avenue, New York, NY 10158-0012

Library of Congress Cataloging-in-Publication Data

Borgaonkar, Digamber S.
Chromosomal variation in man : a catalog of chromosomal variants and anomalies / Digamber S. Borgaonkar. — 7th ed.
p. cm.
Includes bibliographical references and indexes.
ISBN 0-471-02033-8 (alk. paper)
1. Human chromosome abnormalities—Catalogs and collections. 2. Human chromosomes—Catalogs and collections. I. Title.
[DNLM: 1. Chromosome Aberrations—indexes. 2. Chromosome Abnormalities—indexes. ZQS 677 B732c 1994]
RB155.5.B67 1994
616'.042—dc20
DNLM/DLC
for Library of Congress
93-41987
CIP

First edition published by The Johns Hopkins University Press, Baltimore and London, 1975.
Second edition published by Alan R. Liss, Inc., New York, 1977.
Third edition published by Alan R. Liss, Inc., New York, 1980.
Fourth edition published by Alan R. Liss, Inc., New York, 1984.
Fifth edition published by Alan R. Liss, Inc., New York, 1989.
Sixth edition published by Wiley-Liss, Inc., New York, 1991.

The text of this book is printed on acid-free paper.

Carroll, A J, J T Prchal and W H Finley: Transient hematologic disturbance in three members of a family segregating a balanced chromosome translocation. AJHG 36:265(Abstract), 1984; Yang-Feng, T L, G Bruns, A J Carroll, K O J Simola and U Francke: Localization of the LDHA gene to 11p14 to 11p15 by in situ hybridization of an LDHA cDNA probe to two translocations with breakpoints in 11p13. Hum. Genet. 74:331-334, 1986.

Chitayat, D, C L Fagerstrom, D K Kalousek, J Rootman, G P Taylor and J G Hall: De novo reciprocal 1p;2q translocation in a child with multiple congenital anomalies/mental retardation syndrome. AJMG 32:36-41, 1989.
Patinet B.T.
46,XY,t(1;2)(p22;q22).
Some of the features were growth failure, mental retardation, microcephaly, cryptorchidism, partial syndactyly of the second and third toes and an unusual facial appearance.
MIM#270400

de Michelena, M I, J Villacorta and J Chavez: Double chromosome anomaly: interstitial deletion 5q and reciprocal translocation (1;11)(p22;q21). AJMG 36:29-32, 1990.
46,XX,t(1;11)del(5)(pter to q15::q31 to qter)de novo.

Deroover, J, J P Fryns, J Haegeman and H van den Berghe: Paracentric inversion in the short arm of chromosome 1. Hum. Genet. 49:117-121, 1979; Fryns, J P, and H van den Berghe: Paracentric inversion in man: personal experience and review of literature. Hum. Genet. 54:413-416, 1980.
Family V.D.C.
46,XX,inv(1)(p22p36).
Patient MV, 19 years old was an anxious, autistic, severely retarded girl.
46,XY,inv(1)(p22p36).
46,XY,inv(1)(pter to p36::p22 to p36::p22 to qter).

Dhadial, R K, and M F Smith: Terminal 7p deletion and 1;7 translocation associated with craniosynostosis. Hum. Genet. 50:285-289, 1979.
46,XX, t(1;7)(p22;p15), del(7)(pter to p15).
Parental karyotypes were normal.
MIM#218500

Diedrich, U, I Hansmann, D Janke, O Opitz and H D Probeck: Chromosome anomalies in 136 couples with a history of recurrent abortions. Hum. Genet. 65:48-52, 1983.
46,XX,rcp(1;8)(p22;q22).

Francke, U, B de Martinville, L Coussens and A Ullrich: The Human gene for the beta subunit of nerve growth factor is located on the proximal short arm of chromosome 1. Science 222:1248-1251, 1983.

Fryns, J P, A Kleczkowska, E Kubien, P Petit and H Van den Berghe: Cytogenetic survey in couples with recurrent fetal wastage. Hum. Genet. 65:336-354, 1984.
46,XX,t(1;3)(p22;p27).

Fryns, J P, A Kleczkowska, E Kubien and H Van den Berghe: Cytogenetic findings in moderate and severe mental retardation. A study of an institutionalized population of 1991 patients. Acta Paed. Scand. Suppl. 313:1-23, 1984.
46,XY,t(1;9)(p22;p24)mat.
46,XX,inv(1)(p22p25)pat.

Fryns, J P, A Kleczkowska and H Van den Berghe: Paracentric inversions in man. Hum. Genet. 73:205-213, 1986.
Family Nos. 2 and 3.
46,XX, inv(1)(p22p35)/ 47,XX, +21, inv(1)(p22p35)pat.
46,XX,inv(1)(p22q36)pat.

Fukushima, Y, Y Kuroki and T Ito: Balanced double complex translocations [46,XX, t(1p;6p;7p;3q;11p)(11q;22p;21q)] in an infant with multiple congenital anomalies. AJMG 25:313-317, 1986.
Patient (KCMC-90261) was 1 month old.
46,XX, t(1;6;7;11)(11;22;21)(1qter to p22::11p15 to 11pter;6qter to 6p21::1p22 to 1pter;7qter to 7p15::6p21 to 6pter;3pter to 3q27::7p15 to 7pter;3qter to 3q27::11p15 to 11q11::21q11 to 21qter;22qter to 22p11::11q11 to 11qter;21pter to 21q11::22p11 to 22pter).

Geiger, C J, F M Salzano, M S Mattevi, B Erdtmann and F J da Rocha: Chromosome Variation and genetic counseling-20 years of experience in Brazil. Brazil. J. Genet. 10:581-591, 1987.

Case 1789; I. M. F. .
She had 5 miscarriag
karyotypes.
46,XX or XY,t(1;18

Same entry as in 0X
recommendations ha

Kalousek, D K: In
G Wulf: Risks of u
amniocentesis to can
rearrangements: data
laboratories. AJMG
Observation No. 2.
46,XY,t(1;2)(p22;q3
Observation No. 79.
46,XX,t(1;10)(p22;q

McCorquodale, M I
detection of de novo
breakpoints as those
case report. Prenat. I
46,XX,t(1;5)(p22;q2
Normal growth and c

Muro, R, M R Caba
J Egozcue:. A new b
translocation in a ste
1985.
Patient was 42 years
46,XY,t(1;22)(p22;q

Morichon-Delvallez,
Paracentric inversion
observations. Ann. C
Observation 1: M. et
46,XY,inv(1)(p22p3(

Same entry as in 01p

Neu, R L, K Miller
G Wulf: Risks of un
amniocentesis to carr
rearrangements: data
laboratories. AJMG
Observation No. 90.
46,XX,t(1;11)(p22;pl
Observation No. 150.
46,XY,t(1;18)(p22;q2